MT-CYB Gene

Subjects: Genetics
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Definition

mitochondrially encoded cytochrome b

1. Introduction

The *MT-CYB* gene provides instructions for making a protein called cytochrome b. This protein plays a key role in structures called mitochondria, which convert the energy from food into a form that cells can use. Cytochrome b is one of 11 components of a group of proteins called complex III. In mitochondria, complex III performs one step of a process known as oxidative phosphorylation, in which oxygen and simple sugars are used to create adenosine triphosphate (ATP), the cell's main energy source. During oxidative phosphorylation, the protein complexes, including complex III, drive the production of ATP through a step-by-step transfer of negatively charged particles called electrons. Cytochrome b is involved in the transfer of these particles through complex III.

Although most DNA is packaged in chromosomes within the nucleus (nuclear DNA), mitochondria also have a small amount of their own DNA, called mitochondrial DNA (mtDNA). This type of DNA contains many genes essential for normal mitochondrial function. Cytochrome b is the only component of complex III that is produced from a gene found in mitochondrial DNA.

2. Health Conditions Related to Genetic Changes

2.1. Mitochondrial complex III deficiency

Mutations in the *MT-CYB* gene can cause mitochondrial complex III deficiency. When caused by mutations in this gene, the condition is usually characterized by muscle weakness (myopathy) and pain, especially during exercise (exercise intolerance). More severely affected individuals can have problems with other body systems, including the liver, kidneys, heart, and brain.

Most *MT-CYB* gene mutations that cause mitochondrial complex III deficiency change single protein building blocks (amino acids) in the cytochrome b protein or lead to an abnormally short protein. These cytochrome b alterations impair the formation of complex III, severely reducing the complex's activity and oxidative phosphorylation. Researchers believe that impaired oxidative phosphorylation can lead to cell death by reducing the amount of energy available in the cell. It is thought that tissues that require a lot of energy, such as those in the muscles, brain, heart, liver, and kidneys, are most affected by a reduction in oxidative phosphorylation. Damage to these tissues and organs leads to the various features of mitochondrial complex III deficiency.

The location of the *MT-CYB* gene in mitochondrial DNA (mtDNA) may help explain why some people have more severe features of the condition than others. Most of the body's cells contain thousands of mitochondria, each with one or more copies of mitochondrial DNA. These cells can have a mix of mitochondria containing mutated and unmutated DNA (heteroplasmy). The severity of mitochondrial complex III deficiency caused by *MT-CYB* gene mutations is thought to be associated with the percentage of mitochondria with the mitochondrial DNA mutation. In most people with *MT-CYB*-related mitochondrial complex III deficiency, the percentage of mutated mitochondrial DNA is highest in the skeletal muscles, which explains the finding of myopathy in these individuals. It is unclear why the mutation is most prevalent in muscle tissue.

3. Other Names for This Gene

- COB
- CYTB
- cytochrome b
- cytochrome b (mitochondrion) [Homo sapiens]
- MTCYB
- UQCR3

References

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Keywords

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